Renal cystic disease (ADPKD and ARPKD)

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Autosomal dominant polycystic kidney disease (ADPKD) is the most frequent hereditary renal disease and is often encountered in the work-up of renal patients. The diagnosis is obvious in advanced stages, but may be very difficult in young individuals in whom the need to provide a correct diagnosis is particularly pressing.

By ultrasonography cysts are round or oval, echolucent, thin-walled, clearly delineated structures with smooth contours exhibiting sharply demarcated

Fig. 1. (A) 40-year-old male patient, positive family history (mother with ADPKD), hypertension and renal failure. Note numerous large uncomplicated cysts with demonstrable parenchyma in between (right kidney). (B) 56-year-old female patient, positive family history (mother with ADPKD). Note numerous intermediate size echolucent uncomplicated cysts with sound wave amplification behind the cyst in the liver. (C) 17-year-old male patient, positive family history for ADPKD (mother), presenting with borderline hypertension. Note enlarged left kidney with several small echolucent uncomplicated cysts. (D) 30-year-old female patient, negative family history, presenting with renal failure and no liver pathology demonstrable by ultrasonography. Note normal size kidney, echodense parenchyma and scars, small cysts. The patient suffers from ARPKD.

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posterior walls and sound wave amplification behind the cysts as well as lateral extinction of the sound wave (lateral shadowing) [1]. If cysts are solitary or infrequent, they must be distinguished from haematoma or abscess, occasionally also from malignancy, particularly lymphoma.

There are two major types of hereditary polycystic kidney disease, autosomal dominant (ADPKD) and autosomal recessive (ARPKD). ADPKD has three varieties, PKD 1 coded for on chromosome 16, PKD 2 on chromosome 4 and ADPKD 3 on an unknown chromosomal site. ADRKD is coded for on chromosome 6.

The sensitivity of ultrasonography to detect renal cysts in a carrier of the ADPKD trait depends first on the quality of the ultrasonography machine (with outdated machines assessments may not be reliable) and second on the age of the propositus. As a rule of thumb, in individuals aged 30 years, cysts are demonstrable by ultrasound in approximately 68% and above 30 years of age in approximately 89% of carriers of the genetic trait [2].

There are two types of potential misdiagnosis. In very young individuals, the diagnosis may be missed because the renal parenchyma presents with a hyperdense texture only, but even at this early stage hilar and intrarenal vessels exhibit unusually strong vascular reflexes. Occasionally a CT scan permits detection of very small incipient cysts in such cases.

In patients with very large cysts, inexperienced investigators may diagnose urinary tract obstruction. Several patients were referred to us with this misdiagnosis and were shown to have ADPKD. There are some other conditions which may be mistaken for cysts, for example prominent medullary pyramids or megacalycosis (i.e. a malformation and hypoplasia of the papilla with compensatory enlargement of the calyx) [3]. Simple cysts may be difficult to distinguish from ADPKD in young individuals with the ADPKD trait early in the course of the disease when only

![Figure 2](image-url)
isolated cysts are present. As a rule of thumb, the minimum requirement for the diagnosis of ADPKD in individuals less than 30 years of age are two cysts (unilateral or bilateral), at age 30–60 more than a total of five cysts and above age 60 years at least eight cysts bilaterally [4]. The numerous renal cysts in advanced ADPKD may occasionally be difficult to distinguish from acquired cysts in patients with primary renal disease. The most helpful distinctive feature in our experience is the appearance of the renal parenchyma. In the patient with acquired cysts, it tends to be shrunken, whilst in ADPKD one notes harmonic enlargement of the parenchyma interspersed with numerous cysts [3].

Uncomplicated cysts (Figure 1A and 1C) are echo-lucent. Complicated cysts (Figure 2A–D) exhibit a ‘complex pattern’ [1]. This pattern is seen in cases with cyst infection, cyst haemorrhage (with or without cyst rupture) or cyst calcification (usually is a residue after haemorrhage, but the possibility of calcification pointing to renal cell carcinoma should be kept in mind).

In the patient with ADPKD who complains of renal pain with or without macrohaematuria, one has to think of the following complications: cyst rupture, cyst haemorrhage, rupture of a bleeding cyst or bleeding from calyceal varicosity, and rupture of the kidney (after blunt trauma). As soon as the patient has fever or signs of inflammation (e.g. elevated CRP concentration), cyst infection should be suspected, which may require diagnostic puncture of a complicated cyst. Cyst infection is a medical emergency.

Further complications are malignancy (Figure 3A); renal cell carcinoma in a patient with cystic findings should raise suspicion of von Hippel-Lindau disease. In contrast to textbook statements renal cell carcinoma is not more frequent in ADPKD, but does occur. Urinary tract obstruction is a considerate in the differential diagnosis (see above), but large cysts may occasionally cause superimposed obstruction in ADPKD kidneys (Figure 3B) and give risk to kite-like deformation of pyelon and calices (Figure 3C). Renal stone formation (Figure 3D) is more frequent in ADPKD because of stagnant urine, but in the patients
Echodense structures with dorsal shadowing—one must consider cyst wall calcification (after haemorrhage) and renal cell carcinoma (rare).

Table 1 gives some diagnostic clues to distinguish ADPKD (Figure 1A and 1C) from ARPKD (Figure 1D) in early stages of the disease.

To distinguish between the two types, but also to assess the overall risk of the patient with ADPKD, it is useful to be aware of the typical extrarenal findings in ADPKD and ARPKD, respectively. The typical extrarenal abnormalities that can be found by ultrasonography in ADPKD are liver cysts (Figure 1B), pancreatic cysts and rarely cysts of other internal organs. Non-cystic abnormalities associated with ADPKD comprise aneurysm formation of intracerebral arteries and aneurysm formation or dissection of the aorta or other large vessels, diverticulosis of the colon, mitral prolapse (in 25%) or bicuspid aortic valves (by echocardiography) in a sizeable proportion of patients.

In ARPKD one usually finds liver fibrosis with or without portal hypertension and, occasionally, widening of the intrahepatic biliary ducts (Caroli syndrome).

### Teaching point

- In the patient with renal disease and cysts in the kidney, the most important distinction is ADPKD vs acquired cysts in patients with primary renal disease.
- Positive findings which point to ADPKD are a positive family history, relatively well preserved parenchyma between the cysts and extrarenal cysts, particularly hepatic cysts.
- Cyst bleeding and/or infection cause characteristic changes (‘complex pattern’).

### References


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<thead>
<tr>
<th>ADPKD</th>
<th>ARPKD</th>
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<tr>
<td>Normal sized kidney</td>
<td>Enlarged kidney</td>
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<td>Few or multiple usually larger cysts</td>
<td>Multiple small cysts</td>
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<td>Cyst-free parenchyma exhibiting normal echogenicity</td>
<td>Diffuse increase of parenchymal echogenicity</td>
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<td>Medulla clearly distinguishable from cortex</td>
<td>Loss of distinction between medulla and cortex</td>
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